

dbGaP Study Release Notes



Release Notes for NHLBI TOPMed - NHGRI CCDG AFLMU, phs001543.v2.p1

"NHLBI TOPMed - NHGRI CCDG: AF Biobank LMU in the context of the MED Biobank LMU"

For any questions or comments, please contact: dbgap-help@ncbi.nlm.nih.gov.

April 22, 2020 Version 1 Data set release date
July 23, 2021 Version 2 Data set release date

2021-07-23

Version 2 Data set release for NHLBI TOPMed - NHGRI CCDG AFLMU now available

This release includes the addition of Freeze 9 whole genome sequences (WGS) and corresponding VCFs. The study name has been changed from *"NHLBI TOPMed - NHGRI CCDG: AFLMU"* to *"NHLBI TOPMed - NHGRI CCDG: AF Biobank LMU in the context of the MED Biobank LMU."* Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): Health/Medical/Biomedical (IRB, PUB, COL, NPU, MDS) (HMB-IRB-PUB-COL-NPU-MDS)

Data Type	subjects	samples
Phenotype	350	350
Seq_DNA_SNP_CNV(VCFs)	350	350
WGS	350	350

For a description of SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Study and Phenotype Updates

1. New Study Accession

NHLBI TOPMed - NHGRI CCDG AFLMU version 1 phs001543.v1.p1 has been updated to version 2. The dbGaP accession for the current data is **phs001543.v2.p1**. The participant number (p#) has not changed in version 2. No new subjects have been added to this study.

2. There are no updates to the phenotype datasets.

Molecular Data

1. See download components 'sample-info' for manifest of genotyped samples and files.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in 'genotype-qc' tars.

phg	freeze	sample_cnt	subject_cnt
phg001382.v1	8	350	350
phg001606.v1	9	350	350

Authorized Access (Individual Level Data)

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Individual level data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001543/phs001543.v2.p1>

2020-04-22

Version 1 Data set release for NHLBI TOPMed - NHGRI CCDG AFLMU now available

This release includes the addition of Freeze 8 whole genome sequences (WGS) brokered through the Sequence Read Archive (SRA), and VCFs derived from WGS. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): Health/Medical/Biomedical (IRB, PUB, COL, NPU, MDS) (HMB-IRB-PUB-COL-NPU-MDS)

Data Type	subjects	samples
Phenotype	350	350
Seq_DNA_SNP_CNV(VCFs)	350	350
WGS*	350	350

*These data are brokered through the Sequence Read Archive (SRA). Please see Authorized Access instructions below.

For a description of non-SRA SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Molecular Data

1. See download components phg001382.v1.TOPMed_CCDG_AFLMU.sample-info.MULTI.tar for manifest of genotyped samples and files.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in phg001382.v1.TOPMed_CCDG_AFLMU.genotype-qc.MULTI.tar.

Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

dbGaP Study Release Notes



Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001543/phs001543.v1.p1>